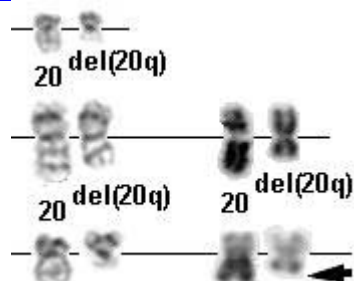


del(20q) in myeloid malignancies

Identity



del(20q) G- banding (left) - Courtesy Diane H. Norback, Eric B. Johnson, Sara Morrison-Delap [Cytogenetics at the Waisman Center](#); R-banding (right) - top: Courtesy Jean-Luc Lai; bottom: Editor

Clinics and Pathology

| | |
|------------------------------|--|
| Disease | a very large spectrum of hematological malignancies as myelodysplastic syndromes (MDS), acute non lymphocytic leukemias (ANLL), polycythemia vera , chronic neutrophilic leukemia |
| Phenotype / cell stem origin | as described in various types of hematological disorders, 20q- appears as a primary karyotypic abnormality occurring in a pluripotential hematopoietic stem cell; the pathogenic mechanism by which 20q- alters the hematopoietic stem cells in hematological disorders remains unknown; 20q- may confer a proliferative advantage to myeloid cells through deletion of a tumor suppressor gene |
| Epidemiology | an interstitial or terminal deletion of the long arm of chromosome 20 (20q-) has been described as the second most frequent sole clonal structural abnormality (5 %) behind t(9.22) |
| Prognosis | <ul style="list-style-type: none"> in MDS, 20q- alone is associated with a good prognosis regarding survival and potential for AML evolution, as defined by the International Prognostic Scoring System (IPSS) for MDS prognosis in de novo acute leukemia, a poor response to treatment and a reduced survival is observed in myeloproliferative disorders, the presence of 20q does not appear to adversely affect survival |

Cytogenetics

Cytogenetics the breakpoint on chromosome 20 is not constant; 20q- is frequently associated with other cytogenetic abnormalities as del(5q), trisomy 8, trisomy 21, deletions or translocations involving the long arm of chromosome 13; a newly described translocation [t\(11;20\)\(p15;q11\)](#) resulting in a [NUP98- TOP1](#) fusion gene was described in therapy-related myelodysplastic syndrome (RAEB); t(11;20)(p15;q11) is a rare recurrent translocation reported in patients with MDS, ANLL and polycythemia vera

Cytogenetics a small fragment (around 8 Mb), proximally flanked by D20S206 and
Molecular distally by D20S119 and UT 654 was identified using FISH
Additional [del\(5q\)](#), [trisomy 8](#), deletions or translocations involving [13q](#) and [trisomy](#)
anomalies [21](#)

Genes involved and Proteins

Note genes remaining within this deleted region are topoisomerase 1 (TPO1-OMIN 126420), phospholipase C (PLC1), hepatocyte factor nuclear 4 (HNF4) and adenosine desaminase (ADA); recently, a new gene KRML transcriptional regulator was mapped in the smallest commonly deleted region in malignant myeloid leukemias

External links

Other database [del\(20q\) in myeloid malignancies](#) [Mitelman database \(CGAP - NCBI\)](#)

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